PHG-1 Public Health Genetics Policy Statement

1.1 Issue

Advances in knowledge and technology related to human genetics are creating opportunities and challenges for public health. An ASTHO policy statement on critical aspects of public health genetics is needed to assist public health agencies with developing their own policies and to communicate shared ideas and positions to policymakers and the public.

1.2 Preamble

State health agencies have been involved with public health genetics through newborn screening programs and maternal and child health activities for over a quarter century. Diseases screened for in these programs are fairly rare, and attention has focused on conditions that are treatable. Today, while discovery of genes related to rare disorders continues, there have been rapid advances in the discovery of genes associated with common diseases, both chronic and infectious. The latter discoveries are creating new opportunities for interventions advancing the health of the public.

Public health program managers and program staff of the future will need appropriate knowledge in genetics to optimally carry out their program mission. For example, breast cancer education programs may be of greater service to women if they incorporate information about genetic mutations associated with breast cancer and are able to make referrals to appropriate genetic services. In the future, it is foreseeable that our understanding of gene-environment interactions will provide more information on disease risk. Eventually, it may be appropriate to recommend avoidance of certain behaviors or environments based on one's genetic profile.

Although knowledge of a person's genetic make-up may be useful in improving health, there are risks associated with determination of an individual's genetic profile. These risks include the possibility of discrimination in employment, insurance, and social interactions, and adverse effects on the mental health of the profiled individual. In several surveys, some participants have indicated that they would not participate in genetic testing because of the potential for genetic information to be used to their detriment by other parties.

The role of public health is to prevent disease and promote the health of the public. Modern genetics presents important opportunities to further this mission. Public health agencies must begin to incorporate genetics into their strategic thinking and consider ways they can take advantage of genetic advances to improve population-based health services.

1.3 General Principles of Public Health Genetics

1.3.1 Assessment: The systematic assessment (surveillance) of factors which are associated with health and disease or with optimal deployment of interventions is well established as a foundation for successful public health programs. As public health agencies expand the

role of genetics in their activities, genetic variables which may become subjects for public health surveillance include: 1) the population frequency of genetic variants that predispose people to specific diseases, both common and rare; 2) the population frequency of morbidity and mortality associated with such diseases; and 3) the prevalence and effects of environmental factors known to interact with given genotypes in producing disease (Khoury et al, 2000). Other important factors include the availability of quality genetic resources in the community, the appropriateness of genetic technologies offered to the community, the accessibility of clinical and genetic services, the cost-benefit of using genetic technology, and the community's knowledge of the use of genetics to improve health status. The assessment of these factors provides a solid foundation for health officials to begin policy and program development.

1.3.2 Policy Development: Genetic technologies can potentially be used in the prevention, diagnosis, and treatment of disease. Public health policies can assist in ensuring that the public can access safe, effective, and quality services without unnecessary apprehension. In addition, public education regarding genetics can facilitate and empower genetic service consumers in decision-making. The development of public health policies regarding genetics should involve the participation of a broad-based spectrum of disciplines, professional backgrounds, interest groups, stakeholders, and consumers. Issues related to genetics which deserve priority policy consideration for policy development are the potential use of genetic information as the basis of discrimination, criteria for and the use of information obtained from population-based genetic screening, privacy and confidentiality of genetic information, including the duty to warn and who should be warned, and protection of the autonomy of individuals relative to genetic profiling and testing.

1.3.3 Assurance: With broad variations from jurisdiction to jurisdiction, public health agencies assure their constituents that necessary health services are provided, either by encouraging action by other private or public sector entities, by requiring such action through regulation, or by providing services directly. Public health agencies can play a constructive role in informing and, where appropriate, encouraging health care providers to incorporate the advances of genetic medicine into their practices. Public health agencies themselves must be alert to opportunities to enhance the fulfillment of program missions by incorporation of techniques and information derived from advances in human genetics. Examples of public health program areas where such opportunities appear to lie include maternal and child health, occupational health, and chronic disease prevention. Enhancement of data systems to include genetic information, with appropriate privacy protections, should be part of ongoing considerations for program improvement. Some health agencies may find it necessary to assure the availability and quality of laboratory and clinical genetics services in their state through licensing and certification activities.

1.4 Specific Positions Statements

1.4.1 Privacy and Confidentiality: The prevention of improper disclosure of medical information is intended to protect individuals from discrimination, to strengthen the health care provider-patient relationship, and to maintain appropriate access to medical information for those conducting medical and public health research. In the past, the

disclosure of genetic information sometimes has led to discrimination and stigmatization. In order for individuals to feel comfortable participating in such research or epidemiological studies, it is essential to protect their confidentiality. Therefore, ASTHO believes that information resulting from medical research and services, including genetics, must be treated confidentially and safeguarded from discriminatory use. The privacy regulation, released in December 2000 as part of the Health Insurance Portability and Accountability Act, provides a basic floor of medical privacy protections for all citizens. In accordance with ASTHO's privacy policy, *PHI. 1. Privacy and Confidentiality of Public Health and Individual Health Data*, states should be able to maintain laws that provide privacy protections more stringent than those offered by federal law.

1.4.2 Genetic Discrimination in Insurance and Employment: According to studies, fear of insurance or employment discrimination may prevent some individuals from participating in genetic testing. Information gleaned from genetic tests can lead to a disease diagnosis, indicate risk of disease, indicate a course of intervention, and provide individuals with information they desire to make life choices. Thus, individuals who avoid testing may miss opportunities to monitor and minimize disease sequelae. Individuals should not be forced to choose between their health and financial security; therefore, ASTHO recommends that all citizens of this country be protected by national policy against discrimination of any form. Each state also should be able to maintain laws that provide protections more stringent than those offered by a national policy.

1.4.3 Informed Consent: A basic tenet of medical ethics is that a person provides his/her informed consent for a procedure. In the case of genetic testing it is critical that informed consent include understanding of the implications of learning and making known one's genetic information. Informed consent for genetic testing should include an explanation of the test's accuracy and meaning of the results, the risks and benefits of learning one's genetic information, other methods for obtaining risk information, and available treatment options. Other than for mandated newborn screening, ASTHO recommends that informed consent be required before any genetic test or screening is performed. ASTHO encourages states to provide parent(s) with information on newborn screening. ASTHO also recommends that states address informed consent for storage and use of genetic material.

1.4.4 Population-Based Genetic Screening: State health agencies have been leaders in population-based screening for conditions which happen to be of genetic etiology for over a quarter century via newborn screening programs. These programs have led to the early diagnosis of mostly rare disorders and, through follow-up and provisions of medical management, prevented unnecessary morbidity and mortality. In addition, these programs have proven to be cost effective by avoiding the expense of lifelong care for a disability. As the genetic nature of common diseases is better understood and the ability to predict the occurrence of these diseases becomes more precise, public health will need to address the integration of genetic testing into screening procedures for common diseases. ASTHO recommends that state health agencies monitor the development of genetic tests and advocate the appropriate use of such tests that can improve population-based screening methods for common diseases. ASTHO also recognizes that genetic

screening should be accompanied with the appropriate education and counseling**,** and that resources will need to be identified to do this.

1.4.5 Public Health Workforce Competencies in Genetics: The integration of genetics into public health will heavily depend on the workforce's ability to envision the impact genetics will have on population-based health and incorporate this into existing and new programs. The Centers for Disease Control and Prevention in collaboration with many stakeholders have developed genetics competencies based on the multiple professional backgrounds and roles of public health practitioners. ASTHO recommends that states determine the appropriate level of genetics competencies for their staff, and that in doing so, the CDC competencies may provide guidance.

1.4.6 Eugenics: In the early to mid twentieth century, approximately 30 states enacted eugenics laws to limit the transmission of perceived undesirable characteristics such as mental retardation by restricting the reproduction of affected individuals. There was little scientific evidence by current standards that would have indicated that a person with one of the targeted traits would have offspring with the same condition; however, tens of thousands of women and men were involuntarily sterilized as a result of these laws. A situation like this must never be allowed to occur.

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